Sheikh Riazuddin

List of Publications by Year
in descending order

103	3,441	172457 29	168389 53
papers	citations	h-index	g-index

103	103	103	5234
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in the Gene Encoding Tight Junction Claudin-14 Cause Autosomal Recessive Deafness DFNB29. Cell, 2001, 104, 165-172.	28.9	430
2	Perrault Syndrome Is Caused by Recessive Mutations in CLPP, Encoding a Mitochondrial ATP-Dependent Chambered Protease. American Journal of Human Genetics, 2013, 92, 605-613.	6.2	186
3	Mutations in the Lysosomal Enzymeâ€"Targeting Pathway and Persistent Stuttering. New England Journal of Medicine, 2010, 362, 677-685.	27.0	181
4	Mutations in FYCO1 Cause Autosomal-Recessive Congenital Cataracts. American Journal of Human Genetics, 2011, 88, 827-838.	6.2	132
5	Dominant modifier DFNM1 suppresses recessive deafness DFNB26. Nature Genetics, 2000, 26, 431-434.	21.4	130
6	Novel mutations of MYO15A associated with profound deafness in consanguineous families and moderately severe hearing loss in a patient with Smith-Magenis syndrome. Human Genetics, 2001, 109, 535-541.	3.8	128
7	Field evaluation and risk assessment of transgenic indica basmati rice. Molecular Breeding, 2004, 13, 301-312.	2.1	103
8	Genomewide Significant Linkage to Stuttering on Chromosome 12. American Journal of Human Genetics, 2005, 76, 647-651.	6.2	100
9	Mutations of Human NARS2, Encoding the Mitochondrial Asparaginyl-tRNA Synthetase, Cause Nonsyndromic Deafness and Leigh Syndrome. PLoS Genetics, 2015, 11, e1005097.	3.5	97
10	Variants in PUS7 Cause Intellectual Disability with Speech Delay, Microcephaly, Short Stature, and Aggressive Behavior. American Journal of Human Genetics, 2018, 103, 1045-1052.	6.2	89
11	A MULTI-CENTER STUDY IN ORDER TO FURTHER DEFINE THE MOLECULAR BASIS OF Î²-THALASSEMIA IN THAILAND, PAKISTAN, SRI LANKA, MAURITIUS, SYRIA, AND INDIA, AND TO DEVELOP A SIMPLE MOLECULAR DIAGNOSTIC STRATEGY BY AMPLIFICATION REFRACTORY MUTATION SYSTEM-POLYMERASE CHAIN REACTION. Hemoglobin, 2001, 25, 397-407.	0.8	73
12	Mutations in TBC1D24, a Gene Associated With Epilepsy, Also Cause Nonsyndromic Deafness DFNB86. American Journal of Human Genetics, 2014, 94, 144-152.	6.2	72
13	A Mutation in SLC24A1 Implicated in Autosomal-Recessive Congenital Stationary Night Blindness. American Journal of Human Genetics, 2010, 87, 523-531.	6.2	67
14	Bi-allelic Variants in METTL5 Cause Autosomal-Recessive Intellectual Disability and Microcephaly. American Journal of Human Genetics, 2019, 105, 869-878.	6.2	58
15	Autosomal Recessive Retinitis Pigmentosa Is Associated with Mutations inRP1in Three Consanguineous Pakistani Families. , 2005, 46, 2264.		50
16	CDC14A phosphatase is essential for hearing and male fertility in mouse and human. Human Molecular Genetics, 2018, 27, 780-798.	2.9	49
17	Global genetic insight contributed by consanguineous Pakistani families segregating hearing loss. Human Mutation, 2019, 40, 53-72.	2.5	48
18	Molecular Genetic Analysis of Pakistani Families With Autosomal Recessive Congenital Cataracts by Homozygosity Screening. , 2017, 58, 2207.		45

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19	A Mutation in ZNF513, a Putative Regulator of Photoreceptor Development, Causes Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 87, 400-409.	6.2	44
20	Mutational and phenotypic spectra of KCNE1 deficiency in Jervell and Langeâ€Nielsen Syndrome and Romanoâ€Ward Syndrome. Human Mutation, 2019, 40, 162-176.	2.5	44
21	Identification of an autosomal recessive stuttering locus on chromosome 3q13.2â€“3q13.33. Human Genetics, 2010, 128, 461-463.	3.8	43
22	Autosomal-Recessive Hearing Impairment Due to Rare Missense Variants within S1PR2. American Journal of Human Genetics, 2016, 98, 331-338.	6.2	43
23	Therapeutic potential of Taraxacum officinale against HCV NS5B polymerase: In-vitro and In silico study. Biomedicine and Pharmacotherapy, 2016, 83, 881-891.	5.6	39
24	Homozygous Truncating Variants in TBC1D23 Cause Pontocerebellar Hypoplasia and Alter Cortical Development. American Journal of Human Genetics, 2017, 101, 428-440.	6.2	39
25	Transformation and inheritance of Bt genes inGossypium hirsutum. Journal of Plant Biology, 2008, 51, 248-254.	2.1	37
26	Mutations in Diphosphoinositol-Pentakisphosphate Kinase PPIP5K2 are associated with hearing loss in human and mouse. PLoS Genetics, 2018, 14, e1007297.	3.5	37
27	Mutations in RLBP1 associated with fundus albipunctatus in consanguineous Pakistani families. British Journal of Ophthalmology, 2011, 95, 1019-1024.	3.9	35
28	Challenges and solutions for gene identification in the presence of familial locus heterogeneity. European Journal of Human Genetics, 2015, 23, 1207-1215.	2.8	35
29	FOXE3 contributes to Peters anomaly through transcriptional regulation of an autophagy-associated protein termed DNAJB1. Nature Communications, 2016, 7, 10953.	12.8	35
30	Homozygosity Mapping and Genetic Analysis of Autosomal Recessive Retinal Dystrophies in 144 Consanguineous Pakistani Families. , 2017, 58, 2218.		34
31	Studies in a consanguineous family reveal a novel locus for stuttering on chromosome 16q. Human Genetics, 2012, 131, 311-313.	3.8	33
32	Investigating the Molecular Basis of Retinal Degeneration in a Familial Cohort of Pakistani Decent by Exome Sequencing. PLoS ONE, 2015, 10, e0136561.	2.5	33
33	Mucolipidosis types II and III and non-syndromic stuttering are associated with different variants in the same genes. European Journal of Human Genetics, 2016, 24, 529-534.	2.8	32
34	Modifier variant of METTL13 suppresses human GAB1â€“associated profound deafness. Journal of Clinical Investigation, 2018, 128, 1509-1522.	8.2	30
35	Missense Mutations in CRYAB Are Liable for Recessive Congenital Cataracts. PLoS ONE, 2015, 10, e0137973.	2.5	29
36	Adipose stem cells differentiated chondrocytes regenerate damaged cartilage in rat model of osteoarthritis. Cell Biology International, 2016, 40, 579-588.	3.0	28

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37	Protective role of vitamin E preconditioning of human dermal fibroblasts against thermal stress in vitro. Life Sciences, 2017, 184, 1-9.	4.3	28
38	CIB2 regulates mTORC1 signaling and is essential for autophagy and visual function. Nature Communications, 2021, 12, 3906.	12.8	28
39	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504.	3.5	25
40	Mutations in the gene encoding the alpha-subunit of rod phosphodiesterase in consanguineous Pakistani families. Molecular Vision, 2006, 12, 1283-91.	1.1	24
41	Loss of function of SVBP leads to autosomal recessive intellectual disability, microcephaly, ataxia, and hypotonia. Genetics in Medicine, 2019, 21, 1790-1796.	2.4	23
42	Studies on the expression of marker genes in chickpea. Plant Cell, Tissue and Organ Culture, 1997, 49, 7-16.	2.3	22
43	Biallelic variants in LINGO1 are associated with autosomal recessive intellectual disability, microcephaly, speech and motor delay. Genetics in Medicine, 2018, 20, 778-784.	2.4	21
44	Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. European Journal of Human Genetics, 2020, 28, 1509-1519.	2.8	21
45	A spectrum of CYP1B1 mutations associated with primary congenital glaucoma in families of Pakistani descent. Human Genome Variation, 2016, 3, 16021.	0.7	20
46	Splice-site mutations identified in PDE6A responsible for retinitis pigmentosa in consanguineous Pakistani families. Molecular Vision, 2015, 21, 871-82.	1.1	20
47	In Vitro Differentiation Potential of Human Placenta Derived Cells into Skin Cells. Stem Cells International, 2015, 2015, 1-11.	2.5	19
48	New insights into Perrault syndrome, a clinically and genetically heterogeneous disorder. Human Genetics, 2022, 141, 805-819.	3.8	19
49	Mutations in the Î²-subunit of rod phosphodiesterase identified in consanguineous Pakistani families with autosomal recessive retinitis pigmentosa. Molecular Vision, 2011, 17, 1373-80.	1.1	19
50	Diazoxide preconditioning of endothelial progenitor cells improves their ability to repair the infarcted myocardium. Cell Biology International, 2015, 39, 1251-1263.	3.0	18
51	Diazoxide preconditioning of endothelial progenitor cells from streptozotocin-induced type 1 diabetic rats improves their ability to repair diabetic cardiomyopathy. Molecular and Cellular Biochemistry, 2015, 410, 267-279.	3.1	18
52	Anti-hepatitis C virus activity and synergistic effect of Nymphaea alba extracts and bioactive constituents in liver infected cells. Microbial Pathogenesis, 2018, 121, 198-209.	2.9	18
53	De novo and bi-allelic variants in AP1G1 cause neurodevelopmental disorder with developmental delay, intellectual disability, and epilepsy. American Journal of Human Genetics, 2021, 108, 1330-1341.	6.2	18
54	Curcumin preconditioning enhances the efficacy of adipose-derived mesenchymal stem cells to accelerate healing of burn wounds. Burns and Trauma, 2021, 9, tkab021.	4.9	18

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55	Mutation in LIM2 Is Responsible for Autosomal Recessive Congenital Cataracts. PLoS ONE, 2016, 11, e0162620.	2.5	17
56	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. European Journal of Human Genetics, 2016, 24, 392-399.	2.8	17
57	Inframe deletion of human ESPN is associated with deafness, vestibulopathy and vision impairment. Journal of Medical Genetics, 2018, 55, 479-488.	3.2	17
58	Autosomal recessive retinitis pigmentosa in a Pakistani family mapped to CNGA1 with identification of a novel mutation. Molecular Vision, 2004, 10, 884-9.	1.1	17
59	Curcumin preconditioned human adipose derived stem cells co-transplanted with platelet rich plasma improve wound healing in diabetic rats. Life Sciences, 2020, 257, 118091.	4.3	16
60	N-Acetyl cysteine protects diabetic mouse derived mesenchymal stem cells from hydrogen-peroxide-induced injury: A novel hypothesis for autologous stem cell transplantation. Journal of the Chinese Medical Association, 2016, 79, 122-129.	1.4	15
61	A genome-wide analysis in consanguineous families reveals new chromosomal loci in specific language impairment (SLI). European Journal of Human Genetics, 2019, 27, 1274-1285.	2.8	13
62	Human neonatal stem cellâ€derived skin substitute improves healing of severe burn wounds in a rat model. Cell Biology International, 2019, 43, 147-157.	3.0	13
63	Deciphering the genetic architecture and ethnographic distribution of IRD in three ethnic populations by whole genome sequence analysis. PLoS Genetics, 2021, 17, e1009848.	3.5	13
64	Mouse Models of Human Pathogenic Variants of TBC1D24 Associated with Non-Syndromic Deafness DFNB86 and DFNA65 and Syndromes Involving Deafness. Genes, 2020, 11, 1122.	2.4	12
65	Vitamin E pretreated Whartonâ€™s jelly-derived mesenchymal stem cells attenuate CCl4-induced hepatocyte injury in vitro and liver fibrosis in vivo. Biochemical Pharmacology, 2021, 186, 114480.	4.4	12
66	Pathogenic mutations in TULP1 responsible for retinitis pigmentosa identified in consanguineous familial cases. Molecular Vision, 2016, 22, 797-815.	1.1	12
67	Serum from CCl₄-induced acute rat injury model induces differentiation of ADSCs towards hepatic cells and reduces liver fibrosis. Growth Factors, 2017, 35, 144-160.	1.7	11
68	Loss of function mutations in RP1 are responsible for retinitis pigmentosa in consanguineous familial cases. Molecular Vision, 2016, 22, 610-25.	1.1	11
69	A mutation in IFT43 causes non-syndromic recessive retinal degeneration. Human Molecular Genetics, 2017, 26, 4741-4751.	2.9	10
70	A novel LRAT mutation affecting splicing in a family with early onset retinitis pigmentosa. Human Genomics, 2018, 12, 35.	2.9	10
71	Standardization of diethylnitrosamine-induced hepatocellular carcinoma rat model with time based molecular assessment. Experimental and Molecular Pathology, 2021, 123, 104715.	2.1	10
72	A Common Ancestral Mutation in CRYBB3 Identified in Multiple Consanguineous Families with Congenital Cataracts. PLoS ONE, 2016, 11, e0157005.	2.5	9

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73	Deletion at the GCNT2 Locus Causes Autosomal Recessive Congenital Cataracts. PLoS ONE, 2016, 11, e0167562.	2.5	9
74	Phenotypic Variability Associated with the D226N Allele of IMPDH1. Ophthalmology, 2015, 122, 429-431.	5.2	8
75	Mutations in phosphodiesterase 6 identified in familial cases of retinitis pigmentosa. Human Genome Variation, 2016, 3, 16036.	0.7	8
76	Vitamin E preconditioning alleviates in vitro thermal stress in cultured human epidermal keratinocytes. Life Sciences, 2019, 239, 116972.	4.3	8
77	Development of <scp>NSAID</scp>â€loaded nanoâ€composite scaffolds for skin tissue engineering applications. Journal of Biomedical Materials Research - Part B Applied Biomaterials, 2020, 108, 3064-3075.	3.4	8
78	In vitro preconditioning of insulin-producing cells with growth factors improves their survival and ability to release insulin. Journal of Biosciences, 2018, 43, 649-659.	1.1	7
79	Mutations in GRM6 identified in consanguineous Pakistani families with congenital stationary night blindness. Molecular Vision, 2015, 21, 1261-71.	1.1	7
80	Mutations in identified in families with congenital cataracts. Molecular Vision, 2020, 26, 334-344.	1.1	7
81	RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. Brain, 2020, 143, e31-e31.	7.6	6
82	A missense allele of PEX5 is responsible for the defective import of PTS2 cargo proteins into peroxisomes. Human Genetics, 2021, 140, 649-666.	3.8	6
83	Biallelic in-frame deletion of SOX4 is associated with developmental delay, hypotonia and intellectual disability. European Journal of Human Genetics, 2022, 30, 243-247.	2.8	6
84	AIPL1 implicated in the pathogenesis of two cases of autosomal recessive retinal degeneration. Molecular Vision, 2014, 20, 1-14.	1.1	6
85	Autosomal recessive congenital cataracts linked to HSF4 in a consanguineous Pakistani family. PLoS ONE, 2019, 14, e0225010.	2.5	5
86	Epigallocatechin-3-gallate protects Wharton's jelly derived mesenchymal stem cells against in vitro heat stress. European Journal of Pharmacology, 2020, 872, 172958.	3.5	5
87	Biallelic variants in TMEM222 cause a new autosomal recessive neurodevelopmental disorder. Genetics in Medicine, 2021, 23, 1246-1254.	2.4	5
88	Novel mutations in RPE65 identified in consanguineous Pakistani families with retinal dystrophy. Molecular Vision, 2013, 19, 1554-64.	1.1	5
89	Are variants in sex hormone metabolizing genes associated with stuttering?. Brain and Language, 2019, 191, 28-30.	1.6	4
90	Antioxidant pretreatment enhances umbilical cord derived stem cells survival in response to thermal stress in vitro. Regenerative Medicine, 2020, 15, 1441-1453.	1.7	4

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91	Novel mutations in identified in familial cases of primary congenital glaucoma. Molecular Vision, 2020, 26, 14-25.	1.1	4
92	Transplantation of stromal-derived factor 1Î± and basic fibroblast growth factor primed insulin-producing cells reverses hyperglycaemia in diabetic rats. Growth Factors, 2017, 35, 88-99.	1.7	3
93	Assessment of potato proteinase inhibitor-II gene as an antifungal and insecticidal agent. Acta Agriculturae Scandinavica - Section B Soil and Plant Science, 2011, 61, 92-96.	0.6	2
94	Identification of Novel Deletions as the Underlying Cause of Retinal Degeneration in Two Pedigrees. Advances in Experimental Medicine and Biology, 2018, 1074, 229-236.	1.6	2
95	Whole genome sequencing data for two individuals of Pakistani descent. Scientific Data, 2018, 5, 180174.	5.3	2
96	Mutations in CERKL and RP1 cause retinitis pigmentosa in Pakistani families. Human Genome Variation, 2020, 7, 14.	0.7	2
97	Alpha lipoic acid priming enhances the hepatoprotective effect of adipose derived stem cells in CCl4 induced hepatic injury in-vitro. European Journal of Pharmacology, 2021, 906, 174201.	3.5	2
98	Comparison of Anti-HCV Activity of Multiple Punica granatum Extracts and Fractions in Virus-infected Human Hepatocytes. Current Pharmaceutical Biotechnology, 2019, 19, 1221-1231.	1.6	2
99	Screening, diagnosis and genetic study of breast cancer patients in Pakistan. Pakistan Journal of Medical Sciences, 2020, 36, 16-20.	0.6	2
100	Effect of age of seedling and phytohormones on micropropagation of indica rice (Oryza sativa L.) from Meristem Culture.. Journal of Plant Biology, 1998, 41, 93-96.	2.1	1
101	Whole-Exome Sequencing Identifies Novel Variants that Co-segregates with Autosomal Recessive Retinal Degeneration in a Pakistani Pedigree. Advances in Experimental Medicine and Biology, 2018, 1074, 219-228.	1.6	1
102	Whole genome sequencing data of multiple individuals of Pakistani descent. Scientific Data, 2020, 7, 350.	5.3	1
103	CLCC1 c. 75C>A Mutation in Pakistani Derived Retinitis Pigmentosa Families Likely Originated With a Single Founder Mutation 2,000â€“5,000 Years Ago. Frontiers in Genetics, 2022, 13, 804924.	2.3	1